

Predictive genetic testing in children and adults: a study of emotional impact

S Michie, M Bobrow, T M Marteau, on behalf of the FAP Collaborative Research Group*

Abstract

Aim—To determine whether, following predictive genetic testing for familial adenomatous polyposis (FAP), children or adults receiving positive results experience clinically significant levels of anxiety or depression, and whether children receiving positive results experience higher levels of anxiety or depression than adults receiving positive results.

Design—Two studies, one cross sectional and one prospective.

Sample—208 unaffected subjects (148 adults and 60 children) at risk for FAP who have undergone genetic testing since 1990.

Main measures—Dependent variables: anxiety, depression; independent variables: test results, demographic measures, psychological resources (optimism, self-esteem).

Results—Study 1. In children receiving positive results, mean scores for anxiety and depression were within the normal range. There was a trend for children receiving positive results to be more anxious and depressed than those receiving negative results. In adults, mean scores for anxiety were within the normal range for those receiving negative results, but were in the clinical range for those receiving positive results, with 43% (95% CI 23-65) of the latter having scores in this range. Regardless of test result, adults were more likely to be clinically anxious if they were low in optimism or self-esteem. Children receiving positive or negative results did not experience greater anxiety or depression than adults. Study 2. For children receiving a positive test result, mean scores for anxiety, depression, and self-esteem were unchanged over the year following the result, while mean anxiety scores decreased and self-esteem increased after receipt of a negative test result over the same period of time.

Conclusion—Children, as a group, did not show clinically significant distress over the first year following predictive genetic testing. Adults were more likely to be clinically anxious if they received a positive result or were low in optimism or self-esteem, with interacting effects. The association between anxiety, self-esteem, and optimism suggests that counselling should be targeted, not only at those with positive test results, but also at those low in psychological resources.

(J Med Genet 2001;38:519-526)

Keywords: genetic testing; children; familial adenomatous polyposis; emotional impact

The increasing availability of predictive genetic testing for late onset diseases means that there is a growing need to understand the psychological consequences of such testing. We know something about the consequences for adults.¹ We know less about groups, such as children, that are regarded as vulnerable to emotional distress and damage to self-esteem.²⁻⁶

In the case of untreatable conditions, the consensus is not to offer children testing because of the possibility of negative emotional consequences and damaged self-esteem for the child.⁵⁻⁶ This is said to result from discrimination, stigmatisation, or altered parental expectations associated with the genetic test results.² The emotional distress experienced by children is said to be greater for children than adults, since they have a longer period in which to live with genetic knowledge and fewer capabilities for dealing with it.⁷

In the case of treatable conditions, the consensus is that children should be offered testing, because the clinical benefits outweigh psychological problems.⁵⁻⁶ Unfortunately, empirical data about the psychological impact of predictive genetic testing in children are scarce.⁸ Such data would be useful to guide clinicians providing predictive testing services for children. A call has recently been made to set up a research protocol to learn more about the impact of predictive genetic testing on children's lives.⁹⁻¹⁰

A recent systematic review of published reports has summarised the psychological impact of predictive genetic testing.¹ Of the 15 papers that met the selection criteria of the review, only one involved children.¹¹ This study assessed 41 6 to 16 year olds by questionnaire before, and three months after, predictive genetic testing for the treatable condition familial adenomatous polyposis (FAP). Children's depression, anxiety, behavioural problems, and competence scores remained in the normal range. The children of affected mothers experienced increases in anxiety and the children of affected mothers with positive test results showed an increase in depression, but these increased levels were within the normal range.

A very different kind of study, a single case interview study of parents before and one month and 15 months after their children underwent genetic testing for FAP, also found no adverse psychological consequences.¹² These parents reported no negative effects on their relationship with their two preschool children and no change in their children's

Psychology and Genetics Research Group, Guy's, King's, and St Thomas's Medical School, King's College London, Guy's Campus, London SE1 9RT, UK
S Michie
T M Marteau

Cambridge Institute of Medical Genetics, University of Cambridge, Wellcome/MRC Building, Addenbrooke's Hospital, Cambridge CB2 2XY, UK
M Bobrow

Correspondence to:
Dr Michie,
susan.michie@kcl.ac.uk

Revised version received
29 March 2001
Accepted for publication
23 May 2001

*D Armstrong, M Bobrow, J Burn, P Chapman, T Clancy, V Collins, D Eccles, G Evans, J Halliday, S Jordan, C McKeown, T M Marteau, S Michie, V Murday, K Neale, R Phillips, J Sampson, J Shea-Simonds, J Weinman

behaviour. They planned to tell their children the results of the testing as and when they asked questions. They felt they were best placed to make this judgement and that parents, not health professionals, should make the decision about whether and when to test children.

Following this study and a review of relevant research,⁸ we set out to describe the emotional impact on children of undergoing predictive genetic testing for a treatable condition, FAP. The typical procedure for such testing is to offer a pre-test visit and a results disclosure visit, with follow up only if needed. We addressed two questions. (1) Do children or adults receiving positive test results experience clinically significant levels of anxiety or depression? (2) Do children receiving positive test results experience higher levels of anxiety or depression than adults receiving positive results?

This paper reports two studies. A cross sectional study compares the psychological impact of predictive genetic testing in children and adults. To the authors' knowledge, this is the first such study. A prospective study compares children's psychological functioning before and at two time points following testing. Because of the small numbers undergoing such testing, these are multicentre studies, involving seven UK genetics centres and one Australian centre.

Study 1. Cross sectional study of adults and children

Methods

The aim was to compare the emotional state of children and adults receiving either positive or negative results following predictive genetic testing, with normative data and with each other. The design is cross sectional.

SAMPLE

The sample comprises adults (aged 17-67 years) and children (aged 10-16 years) at risk of developing FAP who have undergone genetic testing since 1990. None of the adults recruited were parents of the children. They were recruited from seven UK regional genetics centres and one Australian genetics centre. Of the sample of 208, 148 were adults (125 negative and 23 positive) and 60 were children (29 negative and 31 positive). Negative results were "true negatives" in that there was a known mutation in the family. The reason that there were fewer adults with positive compared to negative results is that many of the older adults were, by virtue of still remaining clinically unaffected, at significantly less than 50% risk of having inherited the mutation.

PROCEDURE

Ethical committee approval was obtained for each of the participating centres. Health professionals at the collaborating centres invited those eligible to participate and gave them a study information sheet. People willing to take part were asked to complete a consent form. Parental consent was required for participants aged under 16 years (in one

centre, under 18 years). On receipt of the consent form, the research team sent a questionnaire to the participant, followed by up to two reminders if necessary.

Recruiting clinicians were contacted regularly about the study, by telephone, collaborators' meetings, and visits from the study coordinator. This contact was to monitor recruitment and to ensure that the study protocol was being followed and that no problems had arisen.

MEASURES

The study measures were selected following an interview based pilot study,¹³ with the aim of including well validated measures used in other studies of genetic testing.

Psychological responses

Anxiety. Current levels of anxiety were assessed in adults using the short form of the state scale of the Spielberger State Trait Anxiety Inventory,¹⁴ which was pro-rated to be equivalent to scores obtained using the full form of the scale, giving a range of 20-80 ($\alpha=0.82$). The scale has a cut off of 42, with scores above this signifying clinically significant levels of anxiety. The children's version of the Spielberger State Trait Anxiety Inventory has 20 items and a range of 20-60.¹⁵ Internal consistency was good (α of 0.82 for boys and 0.87 for girls). Norms are available for United States samples: for working adults; they are 35.2 (10.6) for men and 35.2 (10.6) for women, and for children aged 12 years they are 31.8 (SD 5.8) for boys and 30.6 (SD 5.6) for girls. To allow comparison with adult scores, scores on the children's scale were pro-rated to the adult scale range and the adult cut off score was used.

Depression. This was assessed using the Depression subscale of the Hospital Anxiety and Depression Scale, which constitutes seven items asking about feelings in the last week, with four response options per item, giving a range of scores of 0-21 ($\alpha=0.90$).¹⁶ Scores above 7 indicate mild, moderate, or severe depression.

Situational distress. This was assessed using the Impact of Events Scale,¹⁷ comprising seven items measuring intrusion of thoughts about polyposis in the family in the last seven days and eight items measuring avoidance of these thoughts (α for 15 items=0.86). Items were scored 0, 1, 3, or 5, giving a total range of 0-75. The norms for male medical students are 12.7 and 6.9 for female students. For those attending a stress clinic, they are 35.3 for men and 42.1 for women.

Behavioural expression of emotional disturbance (children only). The Rutter Child Behaviour Scale (A) is a brief, well validated scale completed by parents, with a test-retest reliability of 0.74.¹⁹ It has a range of scores of 0-62, with scores below 13 being within the normal range for behavioural problems.

Regrets. Response options for "Do you have any regrets about having the genetic blood test?" were "Yes", "No", or "Don't know".

Perceived threat of test result. This measure was developed from two measures, the Health Orientation Scale (HOS),¹⁸ and a single item of perceived likelihood of getting FAP, ranging from 0 “not at all likely” to 6 “extremely likely”. The HOS was developed to assess the psychological implications of being identified as a gene carrier. It uses the semantic differential technique and asks people to rate their feelings after being given their genetic test result on 10 items: bad-good, vulnerable-safe, risk-not at risk, shocked-relieved, sad-happy, afraid-unafraid, abnormal-normal, sick-healthy, ashamed-unashamed, guilty-not guilty. The scale has a range of 10-50 and an alpha of 0.80 for study data. The combined scale, with perceived likelihood, has an alpha of 0.88.

The HOS and perceived likelihood were combined because factor analysis showed that the HOS loaded highly on a perceived likelihood factor (made up of three other items) and not on a second, emotional factor, made up of anxiety, depression, and situational distress. These two factors were shown to provide a good fit of the data, using maximum likelihood extraction ($\chi^2=7.72$, $df=9$, $p=0.46$).

Perceptions of illness

Perceived chance. Respondents were asked to write a number representing their perceived chance of getting polyposis at some time in their lifetime from 0 “no chance at all” to 100 “absolutely certain”.

Worry about chance. Respondents were asked to rate their worry about chance of getting FAP, from 0 “not at all worried” to 6 “extremely worried”.

Confidence about likelihood. They were also asked to rate their confidence in estimate of likelihood from 0 “not at all confident” to 6 “extremely confident”.

Perceived seriousness. Respondents answered “How serious would you consider the condition if you developed it?” from 0 “not at all serious” to 6 “extremely serious”.

How bad would you feel. Respondents answered “How bad would you feel if you found out that you definitely had FAP?” on a scale from 0 “not at all bad” to 6 “extremely bad”.

Perceived health. Response options for “In general, how would you describe your current health?” were “excellent”, “good”, “fair”, “poor”.

Psychological resources

Dispositional optimism. The two item short form²⁰ of the Life Orientation Test²¹ was used. Five response options were scored 0, 4, 8, 12, and 16, giving a total range of scores of 0-32 ($\alpha=0.76$). The normative score is 21.

Self-esteem. This 10 item scale has a four point scale of agreement, with a range of scores of 10-40 ($\alpha=0.77$).²² It is seen as a personal resource which may moderate the effects of threatening events. The extent to which it reflects a trait or state has not been established. Normative scores are 34.7 for adults and 29.3 for adolescents.

Demographic and test characteristics

The following information was collected from patients: genetic test result (positive, negative), type of genetic test (DNA, linkage), time since testing, age, gender (male, female), and ethnic group (white, non-white). For adults, highest educational qualification (higher than school, school/other, none) and marital status (cohabiting, single/separated/widowed) were recorded.

DATA ANALYSIS

Univariate analyses compared those with positive and negative test results, and adults and children with positive results. Continuous variables were analysed using t tests, apart from chance of getting polyposis which was analysed using Mann-Whitney U, since the distribution was non-normal even after transformation. Categorical variables were analysed using χ^2 tests. The interaction between psychological resource and test result was analysed using a two way analysis of variance.

If less than 30% of data were missing within a questionnaire, the score was pro-rated using the mean replacement method.²³ There were four extreme univariate outliers on time since testing and two on the depression measure, and these were deleted. There were no multivariate outliers. Five variables were non-normally distributed. Time since testing, depression, and child behaviour were transformed by the square root function and confidence about estimate and perceived seriousness were transformed by the logarithmic function.

Predictors of anxiety among adults were assessed using a hierarchical (sequential) regression analysis, which explicitly addresses the issue of correlated independent variables, as was the case in the present study.²⁴ Variables found to correlate with anxiety were entered, with order of entry reflecting a model in which fixed background factors preceded test result which preceded illness perceptions and the threat associated with it.

Results

DEMOGRAPHIC AND TEST CHARACTERISTICS (TABLE 1)

Adults with positive results were younger than those with negative results, which explains the association between cohabiting and negative results. Age and marital status were unrelated to outcomes and so were not included as covariates in the analyses. There was no association between time since testing or age and emotional outcome among either adults or children. Among adults, those receiving positive results were more likely to have undergone linkage testing than those receiving negative results ($\chi^2=7.68$, $df=1$, $p=0.006$). There was no association between type of genetic test and anxiety, so this variable was not included in further analysis.

PSYCHOLOGICAL CHARACTERISTICS (TABLES 2 AND 3)

Children

Depression and anxiety, and its behavioural expression, were in the normal range. Those

Table 1 Demographic and test characteristics of adults and children with negative and positive test results: mean (SD) and frequency

	Test results		Children Negative (n=29)	Positive (n=31)
	Adults Negative (n=125)	Positive (n=23)		
Age	33.9 (12.4)	23.6 (9.4)***	13.7 (1.9)	13.1 (1.9)
Gender				
Male	46 (37%)	10 (43%)	14 (48%)	16 (52%)
Female	79 (63%)	13 (56%)	15 (52%)	15 (48%)
Ethnic group				
White	116 (93%)	21 (91%)	25 (86%)	22 (71%)
Non-white	3 (2%)	1 (4%)	0	0
Missing	6 (5%)	1 (4%)	4 (14%)	9 (29%)
Highest educational qualification				
Higher	36 (29%)	7 (30%)	—	—
School/other	44 (35%)	9 (40%)	—	—
None	13 (10%)	1 (4%)	—	—
Missing	32 (26%)	6 (26%)	—	—
Marital status				
Cohabiting	81 (65%)	7 (39%)***	—	—
Single/sep'd	34 (27%)	15 (65%)	—	—
Missing	10 (8%)	1 (6%)	—	—
Time since tested (weeks)	78.5 (83.2)	115.4 (97.0)†	39.0 (56.7)	53.2 (68.2)
Type of genetic test				
Linkage	22	10**	6	17**
DNA	103	13	23	14

***p<0.001, **p<0.01, *p<0.05, †p<0.10.

receiving negative results had anxiety scores lower than the US norm ($t=4.29$, $df=25$, $p=0.000$). Children receiving positive results perceived their chance of getting FAP as higher, worried more about this, and felt more threatened by their results than children receiving negative results. There were trends towards children with positive results being more anxious and depressed than those with negative results ($t=1.96$, $df=1$, 54 , $p=0.055$ and $t=1.9$, $df=1$, 53 , $p=0.06$ respectively). There was also a trend for a higher proportion of those

receiving positive results, compared to those receiving negative results, to be in the clinical range of anxiety (19%, 95% CI 7-38 v 3%, 95% CI 0.09-18, $p=0.069$, one sided Fisher's exact test). There were no differences in situational distress or behavioural problems between those receiving positive and negative results.

None of the 28 children who received a negative result and three out of 30 receiving a positive result expressed regret at having been tested. Test result did not influence perceived health, with 26 out of 29 negatives and 30 out of 31 positives responding that their health was good or excellent.

Adults

Adults receiving positive results were highly anxious, with mean scores and 43% (95% CI 23-65) of the group in the clinical range. Mean scores were higher both than the norm ($t=2.49$, $df=1$, 20 , $p=0.02$) and than the scores of those receiving negative results ($t=3.49$, $df=1$, 139 , $p=0.001$). Those receiving positive results did not differ from those receiving negative results in regret or in perceived health.

Correlates of anxiety among adults (table 3)

Given the high level of anxiety in adults, a multivariate analysis was carried out to investigate correlates of anxiety in this group. The study variables accounted for 29% of variance. Low optimism and self-esteem explained 14% of the variance of anxiety, with positive test result explaining an additional 12%. After these variables had been entered into the equation, worry about the chance of getting polyposis

Table 2 Psychological measures of adults and children with negative and positive test results: mean (SD) and frequency

	Adults		Children	
	Negative (n=125)	Positive (n=23)	Negative (n=29)	Positive (n=31)
(1) Psychological resources				
Dispositional optimism (0-32)	21.9 (5.8)	21.3 (6.9)	22.2 (6.6)	21.4 (6.1)
Self-esteem (10-40)	33.1 (5.2)	32.7 (5.5)	30.4 (5.5)	32.4 (4.8)
(2) Psychological responses				
Anxiety (20-80)	33.4 (11.1)	43.0 (14.0)***	29.2 (9.1)	33.9 (9.1)†
Children's raw score (20-60)			26.1 (5.9)	29.3 (6.1)
Anxiety "caseness" (adult/child cut off)				
>=42/ >=35	25 (20%)	10 (43%)**	1 (3%)	6 (19%)†
<42/ <35	95 (77%)	11 (48%)	25 (86%)	23 (74%)
Missing	5 (4%)	2 (9%)	3 (10%)	2 (6%)
Depression (0-21)	1.4 (1.8)	1.9 (2.6)	0.7 (1.0)	1.6 (2.1)†
Situational distress (0-75)	14.5 (15.3)	19.7 (16.4)	13.0 (14.2)	15.4 (12.5)
Behaviour scale (0-62)	—	—	9.5 (8.2)	7.7 (3.6)
Perceived threat of test result (11-55)	17.6 (6.2)	35.8 (5.3)***	16.4 (5.7)	32.6 (8.9)***
(3) Perception of illness				
Perceived chance (0-100)	22.9 (30.7)	91.0 (12.4)***	3.5 (7.0)	80.3 (22.6)***
Worry about chance (0-6)	0.9 (1.4)	3.6 (1.9)***	0.7 (1.2)	3.1 (2.1)***
Confident about chance estimate (0-6)	4.9 (1.5)	5.5 (1.0)*	5.3 (1.3)	5.2 (1.4)
Seriousness of polyposis (0-6)	5.4 (0.9)	4.8 (1.4)*	5.4 (0.9)	4.8 (1.6)
How bad would feel (0-6)	4.7 (1.4)	3.8 (1.6)**	4.0 (2.2)	4.1 (1.8)

***p<0.001, **p<0.01, *p<0.05, †p<0.10.

Table 3 Correlates of anxiety among adults: multiple sequential (hierarchical) regression

Step	Variable	Standardised final beta	Raw correlation	R ²	Increase R ²	Partial correlation
1	Optimism	-0.09	-0.31***	0.10	0.10***	-0.09
2	Self-esteem	-0.18	-0.32***	0.13	0.04*	-0.18
3	Test results	-0.16	0.38***	0.23	0.12***	-0.13
4	Worried	0.08	0.40***	0.26	0.04*	0.07
5	Results threat	0.18	0.46***	0.30	0.01	0.12
6	Situation distress	0.19	0.35***	0.33	0.03*	0.20

Anxiety: adjusted R² = 0.29, n=114.

***p<0.001, **p<0.01, *p<0.05.

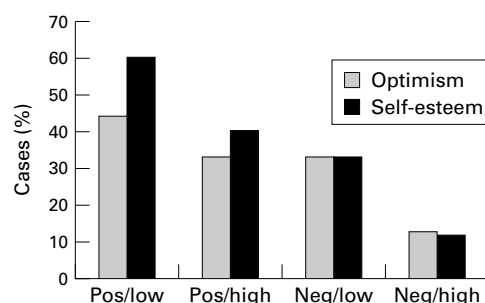


Figure 1 Optimism, self-esteem, and cases of anxiety.

and worry about polyposis in the family explained a further 4% and 3% of variance, respectively. If test result was entered into the regression equation before optimism and self-esteem, test result accounted for 14% of the variance, and optimism and self-esteem accounted for 11% of the variance, showing that test result and psychological resources are independently contributing to the variance in anxiety.

Although both test result and psychological resources were associated with anxiety, they did not interact when anxiety was treated as a continuous variable. However, there were interactions between test result and psychological resource in predicting the frequency of clinical cases of anxiety ($\chi^2=15.80$, df 1, 3, $p=0.001$ for self-esteem and $\chi^2=7.93$, df 1, 3, $p=0.047$ for optimism). Examination of the standardised residuals of the chi-square analyses shows that the greatest contributors to the significant association are the categories of negative test result combined with high self-esteem/optimism (resulting in the lowest levels of anxiety) and of positive test result combined with low self-esteem/optimism (resulting in the highest levels of anxiety). As shown in fig 1, there is the smallest proportion of cases of clinical anxiety when test results are negative and self-esteem and optimism are high (11%, 95% CI 4-22 and 12%, 95% CI 4-24, respectively) and the highest proportion of cases of clinical anxiety when test results are positive and self-esteem and optimism are low (60%, 95% CI 26-88 and 56%, 95% CI 21-86, respectively).

Comparison of children and adults

The only difference between children and adults was that, among those with positive results, children were less anxious than adults ($t=2.6$, df=1, 31.9, $p=0.014$) and fewer children than adults had anxiety scores in the clinical range (19%, 95% CI 7-38 v 43%, 95% CI 23-65) ($\chi^2=4.06$, df=1, $p=0.044$).

Three possible explanations for the difference in anxiety between children and adults receiving positive results are not supported by the data. The first is that children do not understand the meaning of a positive test result. However, children receiving positive results perceive a higher chance of getting polyposis, worry more about that chance, and are more threatened by their test results than children receiving negative results. The second is that they do not perceive polyposis to be as

serious as adults do. There is no difference between children and adults in how serious they consider polyposis to be or how bad they think it would be if they were to develop it. Finally, it may be that high self-esteem or optimism are protective against anxiety. There is no difference between children and adults in either self-esteem or optimism.

Study 2. Prospective study of children

The aim was to examine the course of anxiety in children over the first year after testing. The design is prospective.

Method

SAMPLE

The sample comprised 31 children from the sample in study 1 who had completed additional measures of anxiety before testing and at a second time point after testing. There were no differences in emotional variables or self-esteem after testing between those with and without prospective data. However, the 19 who completed the anxiety measure on all three occasions worried more about FAP in their family at baseline than those who did not ($t=2.9$, df 1, 28, $p=0.008$). There was also a trend for the "completers" to be more anxious at baseline ($t=1.8$, df 1, 29, $p=0.077$) and to be lower in dispositional optimism ($t=2.1$, df 1, 14, $p=0.052$).

MEASURES

The measures were as in study 1. Optimism was not included as data were missing and numbers were small.

PROCEDURE

The procedure was as for study 1, with the difference that three questionnaires were sent to each participant, one before testing and a similar one on two occasions after receiving test results (means of eight weeks, range 1-43, and 33 weeks, range 20-77, respectively).

DATA ANALYSIS

This included non-parametric tests of differences over time and between those receiving positive and negative results. Analyses for each measure were carried out for participants with complete data.

Results

DEMOGRAPHIC AND TEST CHARACTERISTICS (TABLE 4)

Table 4 Demographic and test characteristics of sample ($n=31$): means (SDs)

Age	12.7 (1.9)	Range 10-16 years
Gender		
Male	18	
Female	13	
Ethnic groups		
White	28	
Non-white	4	
Genetic test results		
Positive	16	
Negative	15	
Time since testing (weeks)		
First assessment	8.1 (11.2)	Range 1-43 weeks
Second assessment	32.7 (13.0)	Range 20-77 weeks

Table 5 Means (SDs) of cognitive variables before and on two occasions after receiving test results

			Before: t1	After: t2	After: t3	Within group analyses
Perceived chance	Positive	n=9	53.2 (10.1)	78.9 (25.2)*	72.2 (36.3)†	Friedman $\chi^2=7.5$, p=0.024 Friedman $\chi^2=19.6$, p=0.000
	Negative	n=12	46.3 (13.0)	2.6 (3.9)*	16.2 (22.5)†	
Confidence about estimate	Positive	n=9	4.3 (1.6)	5.2 (1.1)	5.2 (1.1)	Friedman $\chi^2=6.5$, p=0.038 Friedman $\chi^2=11.5$, p=0.003
	Negative	n=10	4.5 (1.1)	5.7 (0.5)	5.7 (0.7)	
Worry about chance	Positive	n=10	3.6 (1.4)	3.4 (1.8)‡	2.1 (1.5)§	Friedman $\chi^2=7.9$, p=0.019 Friedman $\chi^2=15.8$, p=0.000
	Negative	n=12	3.5 (1.6)	0.5 (1.0)‡	0.1 (0.3)§	
Perceived seriousness	Positive	n=10	5.2 (1.2)	4.6 (1.5)	4.5 (1.5)	
	Negative	n=12	5.0 (1.5)	5.2 (1.1)	4.6 (1.6)	
Threat of test result	Positive	n=9	—	27.2 (8.6)¶	29.3 (5.9)**	
	Negative	n=11	—	14.5 (5.7)¶	11.9 (3.3)**	

Between group analyses

*M-W U = 0, p=0.000. †M-W U = 10, p=0.001. ‡M-W U = 11.5, p=0.001. §M-W U = 13.5, p=0.001. ¶M-W U = 7.5, p=0.001. **M-W U = 66.0, p=0.000.

Scores were in the normal range for optimism and self-esteem. Age was not correlated with anxiety.

PSYCHOLOGICAL CHARACTERISTICS (TABLES 5 AND 6)

Positive test results

Children's perceptions of their chance of getting polyposis increased, as did their confidence in their estimate of this chance, after receiving positive results (table 5). They did not, however, show increased emotional distress after receiving positive test results. They worried less about their chance of getting polyposis on the second assessment after testing than previously. Anxiety, depression, and self-esteem scores were within the normal range and did not change over time (table 6).

Negative test results

After receiving negative test results, children's perceptions of their chance of getting polyposis decreased, they became more confident about their estimate of this chance, and worried less about their chance of getting polyposis. They also showed a decrease in anxiety, situational distress, and self-esteem.

Comparison between groups

Children receiving a positive result perceived their chance of getting polyposis as higher, worried more about this, and felt more threatened by their test result at both time points following testing, compared to those receiving a negative result. At the second post-test assessment, those receiving a positive result were

more distressed about FAP in the family and more anxious than were those receiving a negative result. There was no difference between groups in depression at any time point or in self-esteem at the second post-test assessment. It was, however, higher in those receiving positive results at the first post-test assessment, reflecting baseline differences.

Discussion

In children receiving positive results, mean scores for anxiety and depression were within the normal range. There was a trend for children receiving positive results to be more anxious and depressed than those receiving negative results. In adults, mean scores for anxiety were within the normal range for those receiving negative results, but were in the clinical range for those receiving positive results, with 43% having scores in this range. Adults were more likely to be clinically anxious if they were low in optimism or self-esteem. Children receiving positive or negative results did not experience greater anxiety or depression than adults.

CHILDREN'S RESPONSES

One possible explanation for mean scores for anxiety being in the normal range for children who test positive is that this result is based on self-report and children may express their anxiety behaviourally rather than via self-report. This was tested in our study by including a measure of children's behavioural problems; parents' reports did not suggest that they

Table 6 Means (SDs) of emotional variables and self-esteem before and on two occasions after receiving test results

			Before: t1	After: t2	After: t3	Within group analyses
Anxiety (stai-c) (norm 31.0, SD 5.7)	Positive	n=9	32.7 (4.9)	30.6 (6.0)	31.5 (4.2)*	Friedman $\chi^2=12.7$, p=0.002
	Negative	n=9	33.6 (9.0)	25.2 (5.4)	22.8 (3.7)*	
Depression (adult clinical cut-off: 7)	Positive	n=9	2.3 (2.1)	2.6 (3.0)	1.4 (2.3)	
	Negative	n=12	1.4 (1.6)	0.7 (0.9)	0.3 (0.9)	
Situational distress (medical student norms female: 12.7 (10.8), male: 6.9 (6.8))	Positive	n=10	22.1 (8.4)	17.5 (15.2)	15.4 (11.7)†	Friedman $\chi^2=15.8$, p=0.000
	Negative	n=12	20.9 (13.0)	14.5 (13.3)	5.9 (7.4)†	
Self-esteem (norm 29.3)	Positive	n=9	30.7 (5.8)‡	32.2 (4.9)§	32.8 (4.2)	Friedman $\chi^2=11.6$, p=0.003
	Negative	n=11	27.3 (3.4)‡	29.8 (5.5)§	33.5 (6.8)	

Between group analyses

*M-W U = 0.00, p=0.000. †M-W U = 29.0, p=0.023. ‡M-W U = 50.5, p=0.005. §M-W U = 60.5, p=0.029.

had more problems than average. Similarly, in a sample of 41 children undergoing predictive genetic testing for FAP in the United States, anxiety and behavioural problems remained in the normal range three months after testing.¹¹

Mean scores for children's anxiety and depression did not increase over the year following a positive result, despite an increase in their perception of their chance of getting polyposis. Mean anxiety and self-esteem scores decreased after receiving a negative result. Worry about the chance of getting FAP decreased for both those receiving negative and for those receiving positive results.

Although children perceive their risk in accordance with their test result, they are likely to have less understanding of the social implications of the test result, such as obtaining mortgages and insurance, and passing on the condition to children. Alternatively, they may understand the implications but find them less threatening since they are further in the future. However, this is not supported by our finding that children, like adults, find positive results to be more threatening than negative results. It is also not supported by the finding of Codori *et al*¹¹ that among children aged 6 to 16 years, age (taken as a possible marker for comprehension of implications) is not associated with anxiety, depression, or behavioural problems.

Adults show some evidence of threat minimisation,²⁵ since those with positive results perceive polyposis to be a less serious disease than do those with negative results. However, children do not show this pattern, which suggests that threat minimisation is not an explanation for the normal mean score of anxiety in children with positive results. Another possible method of coping is that of regulating a balance between intrusive and avoidant thinking about stressful events. In our study, we referred to this as situational distress, and found no difference between children and adults on this measure. There is a general tendency for people to be unrealistically optimistic about their risk of health problems.²⁶ This has been found for those at high risk²⁷ and for those making estimates about their genetic risk.²⁸ This optimistic bias has been found to be greater among adults than adolescents^{29,30} and it may be that unrealistic optimism leads to an expectation of receiving a negative result and, therefore, raised anxiety on receipt of a positive test result.

Another factor that may explain why children's distress was in the normal range is the support received by children. It may be that children are perceived by health professionals and family members to be more vulnerable than adults and are therefore given more support, both before and after testing. In a recent guide to genetic counselling,³¹ the advice on "giving bad news" includes a section devoted especially to children. The advice (such as structuring information content to the appropriate level, presenting information in clear, simple statements, and repeating it during the session) is advice that would benefit adults as well as children. Children were tested in genetic centres that were sensitive to

children's needs and it is likely that they were given different professional care from that given to adults. Unfortunately, we are not able to assess this from genetic centre protocols, since they are not sufficiently detailed for this. We are currently collecting transcripts of audiotaped disclosure consultations with children and adults to examine communication during the first post-test consultation. Children may also have more support within the family than adults. Research to investigate this has yet to be done.

Two explanations concerning research method may account for the absence of high anxiety among children with a positive result. Although the importance of recruiting all eligible patients to the study was emphasised with our collaborating geneticists, they may have operated different exclusion criteria for children than for adults. They may have used a lower threshold for excluding children from the study who they considered too distressed or vulnerable to participate. Second, the small sample sizes may mean that the study is underpowered. The power of this study to detect the observed effect size of test result on anxiety is 94% in the adult sample and 51% in the child sample.

The prospective data show that emotional state and self-esteem are unchanged after receiving a positive result, while they improve after receiving a negative result. Worry about the chance of developing FAP decreases after receiving a positive as well as after receiving a negative result. This is consistent with an earlier finding among adults that receiving a result, whether positive or negative, resulted in less distress than receiving no result or an uncertain result.³²

ADULTS' RESPONSES

Given that FAP is a treatable condition, it is perhaps surprising that state anxiety levels among adults are so high, higher than those recorded for people with positive results in less treatable conditions, including breast cancer³³ and Huntington's disease.³⁴ It may be that FAP is perceived to be less threatening than breast cancer and Huntington's disease (HD), with the result that those at risk engage less in threat minimising. Alternatively, health professionals may perceive testing for FAP to be less traumatic than testing for hereditary breast and ovarian cancer (HBOC) and HD and therefore provide less counselling. It may also be that those undergoing FAP testing are a less selected, and therefore a less psychologically robust group, than those undergoing HBOC and HD testing. A further explanation is that the high anxiety may be particular to this cohort which has experienced many years of having had the reassurance of negative results following bowel screening. However, this is not supported by the fact that anxiety is no higher for the 117 adults who had undergone previous bowel screening compared to the 23 who had not, and that there is no association between age and anxiety among those who had undergone previous bowel screening.

Among adults undergoing FAP testing, high anxiety was associated with their psychological resources and worries about getting FAP and about FAP in the family, as well as by test results. Adults who received a positive test result and were low in self-esteem or optimism were most likely to experience clinically high levels of anxiety.

STRENGTHS AND LIMITATIONS OF THE STUDY

Study 1 is limited by its cross sectional design, which limits answering questions of causal inference. For example, although the study was based on a psychological model of dispositional optimism as a background variable and general anxiety as an outcome, it may be that anxiety reduces an optimistic outlook, rather than vice versa. Study 2 is limited by the selected sample in that it is of an arguably more vulnerable group of children than the population of children undergoing testing. It may be that those who are less worried and anxious before testing do not show the decreases in worry and anxiety after testing found in this study.

CLINICAL IMPLICATIONS

The role of psychological characteristics in the emotional response to testing found in these studies is consistent with a recent systematic review of the impact of predictive genetic testing.¹ In this review, pre-test psychological functioning was identified as an important determinant of post-test psychological functioning. This suggests that pre- and post-test genetic counselling should be targeted, not just at those with positive test results, but also at those who have fewer psychological resources and who worry more about FAP. It may be that assessing optimism and self-esteem and targeting resources at those low in these psychological resources may help to reduce anxiety. It may also be a more efficient way of using limited counselling resources.

CONCLUDING COMMENT

The results of this study suggest that in the short term at least, there are no adverse psychological consequences for children undergoing predictive testing for FAP when testing is offered as part of a clinical genetics service, which typically includes at least a pre-test and a results disclosure visit. Among adults, receipt of a positive test result is associated with clinically significant levels of anxiety. This is particularly the case among adults with fewer psychological resources. Studies are now needed to determine how counselling is most effectively and efficiently provided to children and adults to achieve good psychological outcomes. In the interim, it would be prudent for those providing services to children to maintain the levels of care and support already in place. The results of the current study, together with the results of the systematic review,¹ suggest that adults with positive test results would benefit from more support and counselling. In addition, pre-test assessment of anxiety and psychological resources could be used as a basis for targeting those most likely to experience high anxiety in the context of predictive testing.

This study was funded as part of a programme grant from The Wellcome Trust. Susan Michie and Theresa Marteau are funded by The Wellcome Trust.

- Broadstock M, Michie S, Marteau TM. The psychological consequences of predictive genetic testing: a systematic review. *Eur J Hum Genet* 2000;8:731-8.
- Clarke A. Introduction. *The genetic testing of children*. Oxford: Bios Scientific Publishers, 1998.
- Grosfeld FJM, Lips CJM, Beemer FA, van Spijker HG, BrouwersSmalbraak GJ, tenKroode HFJ. Psychological risks of genetically testing children for a hereditary cancer syndrome. *Patient Educ Couns* 1997;32:63-7.
- Wertz DC, Fanos JH, Reilly PR. Genetic testing for children and adolescents—who decides? *JAMA* 1994;272:875-81.
- Clinical Genetics Society. *The genetic testing: report of a working party of the Clinical Genetics Society*. Birmingham: Clinical Genetics Society, 1994.
- Reilly PR, Wertz DC. Laboratory policies and practices for the genetic testing of children: a survey of the Helix network. *Am J Hum Genet* 1997;61:1163-8.
- Cohen CB. Moving away from the Huntington's disease paradigm in the predictive genetic testing of children. In: Clarke A, ed. *The genetic testing of children*. Chapter 12. Oxford: Bios Scientific Publishers, 1998.
- Michie S, Marteau TM. Predictive genetic testing in children: the need for psychological research. *Br J Health Psychol* 1996;1:3-14.
- Dickenson DL. Can children and young people consent to be tested for adult onset genetic disorders? *BMJ* 1999;318:1063-5.
- Geller G. Commentary. Weighing burdens and benefits rather than competence. *BMJ* 1999;318:1066.
- Codori AM, Petersen GM, Boyd PA, Brandt J, Giardiello FM. Genetic testing for cancer in children. Short-term psychological effect. *Arch Pediatr Adolesc Med* 1996;150:1131-8.
- Michie S, McDonald V, Bobrow M, McKeown C, Marteau T. Parents' responses to predictive genetic testing in their children: report of a single case study. *J Med Genet* 1996;33:313-18.
- Michie S, McDonald V, Marteau TM. Understanding responses to predictive genetic testing: a grounded theory approach. *Psychol Health* 1996;11:455-70.
- Marteau TM, Bekker H. The development of a six-item short-form of the state scale of the Spielberger state-trait anxiety inventory (STAI). *Br J Clin Psychol* 1992;31:301-6.
- Spielberger CD. *STAIG Preliminary Manual for the State-Trait Inventory for Children*. Palo Alto, California: Consulting Psychologists Press, 1973.
- Zigmond AS, Snaith RP. The Hospital Anxiety and Depression Scale. *Acta Psychiatr Scand* 1983;67:361-70.
- Horowitz M, Wilner N, Alvarez W. Impact of Event Scale: A measure of subjective stress. *Psychosom Med* 1979;41:209-18.
- Wooldridge EQ, Murray RF. The Health Orientation Scale: a measure of feelings about sickle cell trait. *Soc Biol* 1988;35:123-36.
- Elander J, Rutter M. An update of the status of the Rutter Parents' and Teachers' Scales. *Int J Methods Psychiatr Res* 1996;6:63-78.
- Johnston M, Wright S, Weinman J. Individual and demographic differences. In: *Measures in health psychology: a user's portfolio*. Windsor, Berkshire: NFER-Nelson, 1995.
- Scheier MF, Carver CS. Optimism, coping and health: Assessment and implications of generalised outcome expectancies. *Health Psychol* 1985;4:219-47.
- Rosenberg M. *Society and the adolescent self-image* (reprint edition). Middletown, CT: Wesleyan University Press, 1989.
- Afifi AA, Elashoff RM. Missing observations in multivariate statistics. Part 1. Review of the literature. *J Am Statist Assoc* 1966;61:595-604.
- Tabachnick BG, Fidell LS. *Using multivariate statistics*. 3rd ed. New York: Harper Collins, 1996.
- Croyle RT, Yi-Chun Sun, Louie DH. Psychological minimisation of cholesterol test results: moderators of appraisal in college students and community residents. *Health Psychol* 1993;12:503-7.
- Weinstein ND. Unrealistic optimism about susceptibility to health problems. *J Behav Med* 1980;5:441-60.
- Gerrard M, Gibbons FX, Warner TD. Effects of reviewing risk-relevant behaviour on perceived vulnerability among women marines. *Health Psychol* 1991;10:173-9.
- Welkenhuysen M, Evers-Kiebooms G, Decruyenaere M, van den Berghe H. Unrealistic optimism and genetic risk. *Psychol Health* 1996;11:479-92.
- Cohn LD, Macfarlane S, Yanez C. Risk-perception: differences between adolescents and adults. *Health Psychol* 1995;14:217-22.
- Quadrel MJ, Fischhoff B, Davis W. Adolescent (in)vulnerability. *Am Psychol* 1993;8:102-16.
- Baker DL, Schuette JL, Uhlmann WR. *A guide to genetic counseling*. Chichester: Wiley-Liss, 1998.
- Marteau TM, Croyle RT. Psychological responses to genetic testing. *BMJ* 1998;316:693-6.
- Croyle RT, Smith KR, Botkin JR, Baty B, Nash J. Psychological responses to BRCA1 mutation testing: preliminary findings. *Health Psychol* 1997;16:63-72.
- Decruyenaere M, Evers-Kiebooms G, Boogaerts A, Cassiman J-J, Cloostermans T, Demythenaere K, Dom R, Fryns JP, Van den Berghe H. Prediction of psychological functioning one year after the predictive test for Huntington's disease and impact of the test result on reproductive decision making. *J Med Genet* 1996;33:737-43.